

Preface

Since 1992 I have been working in the field of clinical cytogenetics. My diploma, i.e., a master's thesis, was about a special subgroup of patients with small supernumerary marker chromosomes (sSMC), the cat eye syndrome (Liehr et al. 1992). Since that time much progress has been achieved in the field of sSMC. Especially the sSMC homepage (Liehr 2011) with presently more than 4,000 single sSMC case reports together with the advance of technical possibilities for a comprehensive characterization of this special group of rearranged chromosomes enables today much better genotype–phenotype correlations than when I started to study sSMC.

Nonetheless, I recently met a family with the following story, providing evidence that lots of knowledge on sSMC that is nowadays available did not reach the public health system as it should. An sSMC was detected after amniocentesis in the fetus of a pregnant woman who was referred for cytogenetic analysis because of advanced maternal age; sonographic findings were normal. The gynecologist told the couple that the cytogenetic finding was connected with an adverse prognosis and that the developing child would be “100% disabled and mentally retarded.” The parents thus terminated the pregnancy. Later, it turned out that the sSMC was not only parentally derived but also that the first healthy child of the couple also had the same sSMC. This book is intended to help avoid similar situations and to be informative to clinicians, cytogeneticists, and families.

Besides the present knowledge on sSMC, including the biological background, also clinically relevant information is included together with personal reports of families having a child affected with an sSMC. The latter was realized in close collaboration with Unique, the Rare Chromosome Disorder Support Group (<http://www.rarechromo.org/>), and by contributions provided by families in contact with the author.

Jena, October 2011

Thomas Liehr

References

- Liehr T (2011) The sSMC homepage. <http://www.med.uni-jena.de/fish/sSMC/00START.htm>. Accessed 10 Jan 2011, Also accessible via <http://markerchromosomes.wg.am> or <http://markerchromosomes.ag.vu>
- Liehr T, Pfeiffer RA, Trautmann U (1992) Typical and partial cat eye syndrome: identification of the marker chromosome by FISH. Clin Genet 42:91–96

Small Supernumerary Marker Chromosomes (sSMC)

A Guide for Human Geneticists and Clinicians

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